TWO CASES OF FRIEDREICH'S DISEASE.*

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I ESPECIALLY desire in this paper to report two cases, which I consider clinical types of that interesting form of hereditary degeneration of the spinal cord designated as Friedreich's disease.

I select these two, because they differ in many points clinically, though they both serve to illustrate some interesting points for consideration as to the pathological nature and extent of the disease.

Miss H., age twenty-nine.

Family History.—Both parents living. The father is of the neurotic type. Emotional; delicate. Discouraged easily and often has broken down under business cares.

The mother is also of the emotional neurotic type and suffers from migraine. She has a brother in the asylum. A sister with narcomania, and another brother of very eccentric habits and mental peculiarities. There were six children.

The oldest is the patient, Miss H.

The next is a man of twenty-seven, very much undersized, and of delicate physique.

The next is a man of twenty-five, in good health.

The fourth is a girl of nineteen, very much undersized, and in appearance and actions undeveloped; suffers from migraine. Has menstruated from thirteenth year of her age. Menstruation has been irregular. At times profuse, at others scanty. At times appearing two and three weeks apart; again not appearing for two months. Suffers from profuse nose bleeds. Has a small undeveloped uterus.

The fifth child, a boy, was delicate from childhood. Very nervous and slightly choreic—in fact, lived with difficulty to the age of sixteen, and died with a particularly short attack of pernicious anæmia.

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The sixth child is a robust boy—developing into a apparently strong manhood.

Personal History.—The early history of the patient was uneventful to near the age of puberty. She had then attacks of supposed malaria. She menstruated at thirteen. At this time she consulted the late Dr. Knight for being very round-shouldered and ungraceful in her movements. He diagnosed double lateral curvature. The patient wore a steel corset for one year, night and day, and another year during the day only. The curvature, however, increased, and she grew more round-shouldered and ungraceful.

In August, 1876, age fifteen, she had an attack of supposed rheumatism ; no swelling ; only pain in the joints of the extremities.

In spring of 1878, there was so much headache, backache, and erratic pains in the extremities that the girl was obliged to leave school. At this period ungracefulness led to frequent falls, and she was still supposed by physicians to be suffering from rheumatism. The patient had a number of attacks of pain in head, back and extremities, each lasting from two to three weeks and each followed by more than the usual ungraceful gait up to 1882, when a more than usual severe attack of headache and backache left her unable to walk without assistance or stand without swaying, and if it was attempted with eyes closed she was liable to fall. She had suffered since puberty, up this date, with attacks of cramps in the calves of her legs, mostly occurring at night, very soon after she fell asleep.

Patient talked badly and had a disagreeable eruption on knees, elbows and knuckles. Elevated papules, capped by scales. This eruption lasted two years and slowly disappeared. She consulted Dr. L. A. Sayre in 1882, who treated her double lateral curvature by extension and placed her in a plaster jacket. The patient, however, grew rapidly ataxic. In 1883 she suffered from profuse and frequent menstruation, being obliged to arise as often as twelve times at night. She was unable to perform fine coördinating movements with the hands. Her facial expression was unmanageable. The muscular movements were overdone. For instance, a smile was a grimace; an attempt to wink caused a grotesque facial spasm.

The slurring and rapidity, with ommission of labials, made speech perplexing.

Nystagmus was an early symptom, but the exact time it was first noticed is not remembered. It was at first most constantly present when the patient was about sixteen years of age. The patient has suffered more or less constantly with erratic pain in head, back and extremities.

I first saw patient April 7, 1884. She is still under my professional care.

Examination.—A handsome girl; blonde, slight and small of stature; well-nourished; intellectually bright; no mental derangement. Patient had a marked ataxic gait, unable to walk or stand alone. Sitting, patient exhibited marked ataxia of body muscle, ataxia of upper extremities, face and speech muscles. All muscular movements awkward and overdone. Tactile sense good. With æthesiometer variable and slight anæsthesia noted. Poor weight sense. Heat and cold were easily distinguished. Abolition of the deep and superficial reflexes. Electrical examination negative. Ophthalmoscopic examination negative. No pupillary symptoms. The double lateral rotary curvature of the spine noted.

Summary to Date.—The patient has had variable attacks of headache and backache. Vaso-motor troubles noted in irregular action of heart at times. Flushings of face, extremely cold hands and feet, with chilblains in winter. Polyuria at times. Attacks of pulsations in groins and vulva, with heat and swelling of the latter, lasting days. Ataxia increased slowly, especially of the lower extremities. Muscular strength fair. The patient has preferred to remain in bed most of the time of late. There is no muscular weakness sufficient to absolutely confirm a paresis supervening.

This is undoubtedly a case of early systematic degeneration of the posterior columns, extending to medulla and various vaso-motor centres.

The prominent clinical features, tabulated as follows, are to be seen in typical cases of Friedreich's disease:

1. The age of development.
2. A general ataxia.
3. No fixed anæsthesia.
4. Nystagmus.
5. Ataxic speech.
6. Abolition of reflexes.
7. Curvature of spine.
8. Vaso-motor symptoms.

This group of symptoms form the most general ones noted in this class of cases.

The atypical or anomalous symptoms are:

1. Initial pain.
2. Spasms.
3. No apparent essential paralysis.

The initiatory attacks of pain, however, lacked the character of true lancinating pain of tabes. They were more constant, and confined to head, back and joints; and this character of pain has been often recorded in cases of Freidreich's disease. Griffith found these initial pains in 22 of the 143 reported cases he had analyzed.

The cases reported by Mussa, Botkin and Friedreich, all suffered from initial headache. Bradbury reports a case with pain and weakness of the back.

One of Carres most typical cases suffered with severe initial pain in legs and feet. Pain concomitant with the disease, and after it is established is not so uncommon.

Forty-five cases tabulated by Griffith had neuralgic pains of various nature. The attacks of pains in my case were more frequent after the disease was noted and well established. Pain is merely anomalous symptom, inasmuch as it is not a precursor of Friedreich's disease with that severity as noted in tabes.

What causes the pain in tabes has been a question for much discussion. Meningitis, degeneration of posterior roots, or peripheral neuritis. This is uncertain as yet, though degeneration of posterior roots is conceded the chief cause.

Why the pains of ataxia should exceed those of Friedreichs' disease is of interest. Déjerine, in the discussion on the nature of the sclerosis of Friedreichs' disease before the Société de Biologie, March 7, 1880, claimed that the lesser difference in degree and character of the pain, in the disease under consideration than in tabes, is not due to unequal or less alteration in the posterior roots, for in the cases under his observations the posterior roots were as much diseased as in tabes. But the fact of a difference in the character of the degeneration might explain why we are more apt to have severer pain with tabes than in Friedreich's disease.

In the former we have the degeneration closely allied to an inflammatory process, while in the latter we have a disease undoubtedly depending upon an evolutionary process.

It is easy to understand that, in the breaking down of young and most delicate tissue less resistance is met with and, consequently, less response in functional disturbances. In this case of mine pain was a factor, and the question naturally suggests itself, was it not due to a more active disturbance in the breaking down of older and therefore stronger and more resisting nerve tissue, and to deduce the inference that had the degeneration occurred at an infantile period than at puberty and adolescence, that the process of dissolution would not have been accompanied by pain, or at least not much.

It is natural to surmise that at puberty more active and irregular disorders of functions are apt to occur and all symptoms to assume a more active type.

One of the interesting features of this case is regular occurrence of muscular spasms at night. This is not of unfrequent occurrence in the reported cases; but I will leave its discussion till I take up some analogous features with the next case.

Muscular weakness would naturally be expected to be present more or less in a case of advanced ataxia.

In the reported cases of Friedreich's disease this symptom has not been specially spoken of in many cases. The paralysis was not considered an important feature, and the inability to walk was not in all cases related to any degree of paresis present. Griffith gives 56 cases as pronounced ones exhibiting more or less muscular paralysis. Again, many cases reported were in young cases where the disease was not advanced. Whether the inability to walk in time will result in secondary complications and degenerations of the lateral tract is a question.

The case I report is certainly far advanced in her ataxia, and has for years been unable to walk much or has used her limbs to any extent, yet muscular weakness is not pronounced and a true paresis is not present, and whether the case develops essential paralysis certainly is a question.

The following case has been described by Dr. Dana,[1] in an article entitled "Combined Sclerosis of the Spinal Cord." His description was complete, but differs slightly in a few particulars from that obtained by myself:

Jane D., aged twenty-eight; single; no occupation.

Family History.—The grandmother (father's side) had epilepsy. The father, when a young man, drank, and suffered from frequent "nervous spells" with vertigo, and always complained of indigestion, and has doctored ever since he could remember. The mother had been physically a weak woman many years before this child was born. She has had three young children die in convulsions at the ages respectively of sixteen, eleven, and eight months. She has three grown daughters, all of whom were very nervous as children. One, now married, is reported to be in fair health. Another, aged twenty-one, has often complained of having strange feelings and thoughts with uncontrollable morbid impulses to run and shout; suffers from attacks of migraine, and is hysterical from any slight physical or mental strain.

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[1] Medical Record, July 2, 1887.

Personal History.—As long as the mother can remember the patient was a nervous child. She had no physical endurance, tired easily; she had frequent attacks of mental confusion, imagining she was in other places, with complete loss of personal identity or those of friends and relatives that might be around her. These attacks evidently were epileptoid.

From description patient had attacks of psychical blindness. Troubles with vision were frequent, and, as she grew toward the age of puberty, she became more disinclined to exert herself. She menstruated at fourteen, and has been regular, though the flow always was very scanty. No history of genito-urinary troubles.

She has lived a quiet, uneventful life, without vicious habits. As her disease progressed, she has shown less and less hysterical and epileptoid symptoms. The symptoms were prominently apparent when about sixteen. She was obliged to look where she was going or she would be apt to fall; and she walked badly, and stumbles and falls were frequent. This was followed in a year by numbness and tinglings, with some weakness, and the numbness extended to the hands, more in the right than the left.

In 1884 these symptoms were constant, with considerable weakness in the legs. From this time symptoms became rapidly worse, with coldness of extremities, slight puffiness of feet, formications, and slight delay in act of micturition. Her feet felt like rubber balls on the floor; she staggered when she walked, and the heels struck the floor first.

At this time inco-ordination was the marked symptom, though she was weak and limbs were slightly stiff, with occasional cramps in muscles; but she never had acute pain. She could not use her hands for delicate co-ordinating work, such as sewing: the needle was not felt and held securely without the aid of vision, and great labor was necessary to make the stitches even.

1885 and 1886.—She grew worse, and became unable to walk. Stiffness with cramps was present a great part of the time, but diminished the latter part of 1886, and continued to do so up to my first seeing the case in the early part of 1889.

Examination.—She was well nourished; complained of not being able to walk or to use her hands freely. By assistance could take a few steps; at times able to go about the room with her sister's aid. Limbs not stiff; ankle-clonus elicited as well as tendo-reflex; more marked on re-enforcement.

Slight anæsthesia of both lower extremities, with slight hyperæsthesia of both plantar surfaces. Heat sense good; weight and space sense poor.

In the upper extremities muscular weakness marked; more noticeable on right side; right arm feels also more numb. No reflexes obtainable; slight anæsthesia: heat sense good; weight sense and space sense poor.

The extremities always feel very cold, and are abnormally so. Electrical examination negative; ophthalmoscopic examination negative; pupillary reflexes normal; no nystagmus; no curvature of spine; speech is slow, but not ataxic. Patient is eccentric, stubborn, and slovenly.

January 1, 1890.—Patient suffered from a sudden attack of diplopia (right sixth nerve paralysis), with drawing of muscles of left side of the face and paresis of the right side, and vomiting. This was a probable central hemorrhage, and will not be further considered in the discussion, except to say it is now hardly discernible.

This case of early progressive degeneration of the cord presented first marked ataxic symptoms, followed by ataxic paraplegic condition, with spastic symptoms for a time.

The anomalous symptoms are the quickly developing paresis with spastic symptoms. The presence of the reflexes and ankle-clonus are difficult to explain, unless we accept Westphal's explanation in like cases: that knee-jerk will not be abolished unless the lumbar enlargement be involved. And Griffith, in his summary, concludes that genuine cases may occur in which the reflex are slight, normal, or even exaggerated.

In twelve cases reported the reflexes were present; in six exaggerated. Spasm of muscles occurred in twenty-one cases, and marked ankle-clonus in two cases.

There is no marked spastic condition in any of the muscles of the lower extremities in this case now. This was a symptom occurring in the more active stage of the disease.

Strumpell speaks of the hereditary nature and family type in spastic ataxia, and one he reports[2] which resembles mine in many particulars. Dr. Dana speaks in his article of the importance of heredity as a factor.

In spite of the quickly developing paresis and the presence of the reflexes with spasm, I think this case must be classed in the category of the hereditary degenerations. It is probable that the degenerations occurring in the various tracts in both these cases were due to evolution, the cord in part being predestined to early destruction.

In many cases of Friedreich's disease it is conceded that it is not necessary to have an exact type of the disease in a parent or ancestor or even a correlation with a neurosis or neuropathic taint. It has been found, however, that the 143 cases reported by Griffith occurred in 77 families.

There has been an endeavor on the part of investigators to seek for the family type, and more attention has been paid to the reporting of these than to the isolated cases. This undoubtedly leads many to look upon the isolated cases as not typical ones of Friedreich's disease, and especially if they in any way present anomalous symptoms.

Many just such cases as those I report, which differ markedly from the acute degenerations of the adult, and yet not directly traceable to heredity, point strongly to anomalies of development of probably ectodermic origin.

To lay too much stress upon the imperfect nervous mechanism of ancestor or parent as the cause in these cases is not practically wise, and is apt to draw attention away from the consideration of other unknown (perhaps physical) forces at play in the vital structure of the blastoderm.

It is of great scientific value to study, as near as possible, the embryonic origin of the tissue itself, which undergoes a retrograde change, and the character of the pathological evolution.

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[2] Arch. f. Psych., vol. xvii., No. 1, case iii.

The importance of histogenesis of tissue to pathological study is becoming more apparent. Blastodermic derivation of tissue is being carefully investigated in new ways, as from the side of morbid anatomy. Formad[3] demonstrates that the derivation of the tissue determines the variety of new growths in that tissue and their character of development, so that prognosis and diagnosis can readily be made. The character of the pathological evolution of tissue is toward that of the early derivative tissue.

Chaslin demonstrated that in epilepsy there exist cortical gliomata in the form of fibrillary tissue analogous to that of the neuroglia, without any vascular lesion or sclerosis.

Déjerine and Letulle, in a paper on the nature of the sclerosis in Friedreichs disease,[4] discuss the histological differences between this form of degeneration and that of tabes or other sclerosis. In Letulle's case the degenerative changes were of the neuroglic character.

Tangled fibres of various lengths and every possible arrangement. Nothing of note was to be found in connective-tissue change in blood-vessels, except slight hyaline degeneration of the lymphatic sheath of the capillaries. Prolongation of pia mater was normal and intact; this, however, was chiefly located in the posterior columns: more positive evidences of connective-tissue sclerosis of a vascular origin being found in the lateral columns and other tracts.

Gliomatous masses were seen as well about the central canal. This gliomatous histological nature of sclerosis in Friedreich's disease unmistakably points to anomalies in development and the evolution toward derivative neuroglic tissue.

The evolution in Friedreich's disease follows more or less a regular order, with undoubtedly a tendency to implicate the entire cord. It insidiously selects the posterior columns first, creeping on and spreading out into other portions slowly. There is undoubtedly a more active stage, which completes more quickly and for all time the gross extent of the lesion, and the disease again halts and the degeneration progresses more slowly. In my first case remissions and exacerbations were marked and extended over several years.

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[3] University Med. Mag., May, 1890.

[4] Le Médecine Moderne, March, 1890.

The implication of all parts of the cord in the degenerative process has lately been observed by Blocq and Marinescu.[5] In abstract it is as follows:

Considerable diminution in volume of the entire cord, which is abnormally small in any given area. The entire columns of Goll are affected throughout—the column of Burdach irregularly—as far as the decussation of the pyramids.

The crossed pyramidal tracts are entirely changed, the lesion decreasing from below upward as far as the decussation. The cerebellar tracts are affected from the lower dorsal region.

Gower's tract is not abnormal within this region. Lissauer's zone is the seat of disease in lower and middle lumbar region. The marginal external tract is unaffected. The columns of Clark are profoundly changed throughout their entire length. Histological lesions occur even when there is no appearance of gross abnormality.

Fewer large healthy fibres than there should be are present; and there is an alteration in their disposition. There were changes in the funiculi gracilis, the cerebellar tracts, and the funiculus cuneatus of the medulla. Besides these various degenerations, there was vascular dilatation of lacunæ form existing throughout the changed tissue. *In toto* an entire implication of the cord and a destructive degeneration of a great portion.

The stereotyped clinical phenomenon observed in Friedreich's disease is that of ataxia with more or less paralysis, and these complicated irregularly by myopathies, deformities, vaso-motor derangements, and even cerebral difficulties.

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[5] Le Progrès Médical, March 8, 1890.

The variations in the ataxia relatively to the degree and extent of the paralysis, and more or less prominence at times of the complications, leads one to classify cases often as types of ataxic paraplegia, spastic paraplegia with some ataxia, or again perhaps as a case of imbecility with ataxic paraplegic complications.

To classify the variations of Friedreich's disease into types will not aid in the better understanding of the true nature of the disease, and will not be satisfactory, as no two cases are exact in various clinical phenomena. Anomalies in the clinical picturing are to be expected, but it will not be difficult to adjust these when we recognize the nature of the pathological process and its rather extensive irregular distribution without fixed limitation to the posterior columns or even posterior columns and lateral tracts.

THE PARALYSES OF LARYNGEAL MUSCLES DURING THE COURSE OF TABES DORSALIS.

Dr. R. Dreyfuss (Virchow's Archiv, 1890, Bd. 120, p. 154), after a historical review, gives the results of his investigations, made in Mendel's Polyclinic. There were twenty-two patients in all. He found in two patients paralysis of the posterior crico-arytenoid muscle in the first stage of development. In the other patients laryngoscopic examination did not reveal any motor disturbances which could be said to be due to tabes; in all the cases there was a total lack of change in sensation. The writer then gives the description of three other cases of tabic paralysis of the vocal cords.

He objects to the use of the term "real ataxia" of the vocal cords in tabes, proposed by Krause, for one can only speak of ataxia of a group of muscles; but on inspiration only one muscle in the larynx, the posterior crico-arytenoideus, comes into action. The cases of phonetoric ataxia of tabes described up to now only present this disturbance of co-ordination as transient; hence this symptom is not persistent. He does not by any means consider tabic laryngeal affections as frequent as Krause (thirty-three per cent.) and Marina (fifty per cent.).